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Metatropic dysplasia – case reports

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Summary

Background:

Metatropic dysplasia (changeable dysplasia) presents with characteristic clinical and diagnostic radiographic findings already present at birth. The aim of this paper is to familiarize radiologists, pediatricians and orthopedic surgeons with this relatively common bone dysplasia and to stress the importance of its early diagnosis.

Case Reports:

Six patients with Metatropic Dysplasia and one with Metatropic Dysplasia Variant are reported. The group consisted of 6 males and 1 female.

Conclusions:

Early diagnosis of Metatropic Dysplasia is of utmost importance for appropriate treatment and prognostication of this disorder, apparently "benign" at birth. Late diagnosis is the result of unfamiliarity with this bone dysplasia,

Key words:

metatropic dysplasia • bone dysplasia • scoliosis • kyphoscoliosis

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Background

Metatropic dysplasia (MD) (OMIM 156530 and 250600) is a relatively common spondyloepimetaphyseal dysplasia with characteristic clinical and diagnostic radiographic findings [1–5].

Early diagnosis is of utmost importance for appropriate prognostication and treatment. For the researcher unfamiliar with the disease, the appearances of a newborn with pleasant face, slightly long trunk and shortened extremities does not foreshadow a disorder with progressive serious clinical course and often early fatal outcome. Progressive kyphoscoliosis in spite of early treatment implicates the small thorax with vulnerability to early pulmonary complications.

Case Reports

For clarity, we present the pertinent clinical data important for the diagnosis and treatment as a summary in a Table 1.

MD – a disease of considerable variability – is of interest to radiologists, pediatricians and orthopaedic surgeons. Some patients are born dead or die soon after birth. Most perish in the first two decades of life. In rare instances survival to adulthood occurs. All our children had negative family history, normal mental development and normal routine biochemistry including Ca/P balance.

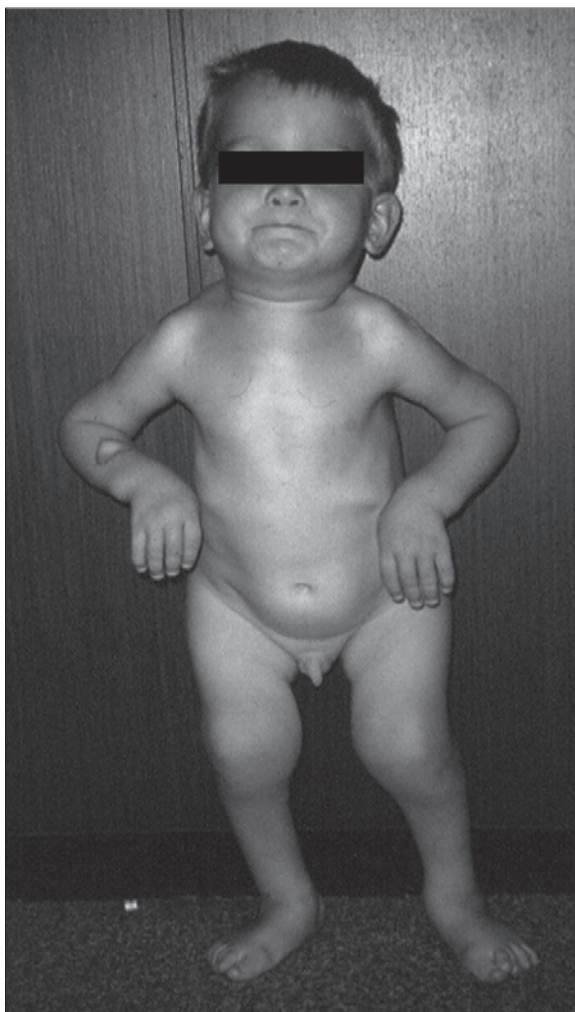
Discussion

MD is usually easy to diagnose as it demonstrates characteristic clinical and diagnostic radiographic features.

The major clinical finding at birth is disproportion between the long trunk and the short extremities. A face without dysmorphic features, and a tail-like appendage in the sacral region are two important additional landmarks. The alarming signs which usually appear in the first year of life are delayed motor development, swelling and contractures of the joints and kyphoscoliosis with chest deformity. Progressive kyphoscoliosis leads

Table 1. Table demonstrates late diagnosis of Metatropic Dysplasia in spite of early dysmorphic clinical and radiological features.

Birth weight, length, sex	Abnormalities noted at birth	Kyphoscoliosis noted at age	Dg of MD	Remarks
1 2220 g, 48 cm, M	None	1month	3 y	Bone dysplasia diagnosed at 1 month; At 13 y 128 cm (–3.5SD); Wheel chaired
2 3750 g >50 cm, M	Tail-like appendage	After birth	1 m	Rapidly progressing kyphoscoliosis; At 13 m 76 cm (–1SD)
3 3260 g, >50 cm, M	“Deformities of the skeleton”	During 1 st year of life	12 m	Progressive big joints contractures; At 4 y 79 cm (–6.0SD); Wheel chaired
4 3750 g, 50 cm, M	Short extremities	During 1 st year of life	19 y	Chondrodystrophy dg at 1 y. Kniest disease at 12 y; Wheel chaired; At 15 y 123 cm (–5.5SD)
5 3500 g, >50 cm, F	None	During 1 st year of life	4.5 y	Tracheomalacia. At 4.5 y 86.5 cm (–4.0SD); Wheel chaired
6 3525 g, 49 cm, M	Prominent big joints with contractures	During 1 st year of life	17 m	At 7.5y 107.4 cm (–3.5SD)
7 ?	?	?	3 m	Single consultation X-ray send without any clinical data!

**Figure 1.** MD Case 1 7y old boy. Short trunk. Swelling and contractures of the big joints. Normal skull and face.**Figure 2.** MD CASE 5. 21y old woman. Severe kyphoscoliosis.

to change of phenotype – short trunk/long extremities as reflected in the name of the disease – metatropic = changing (Figures 1,2). The mental development of the children is normal. In our Patients 2 and 3 kyphoscoliosis was present already at birth and it progressed rapidly during the first year of life (Figure 3A). Early diagnosis is of utmost importance for proper management and prognostication. Parents should be informed of the severity of the disease in spite of the benign characteristics of the disease at birth.

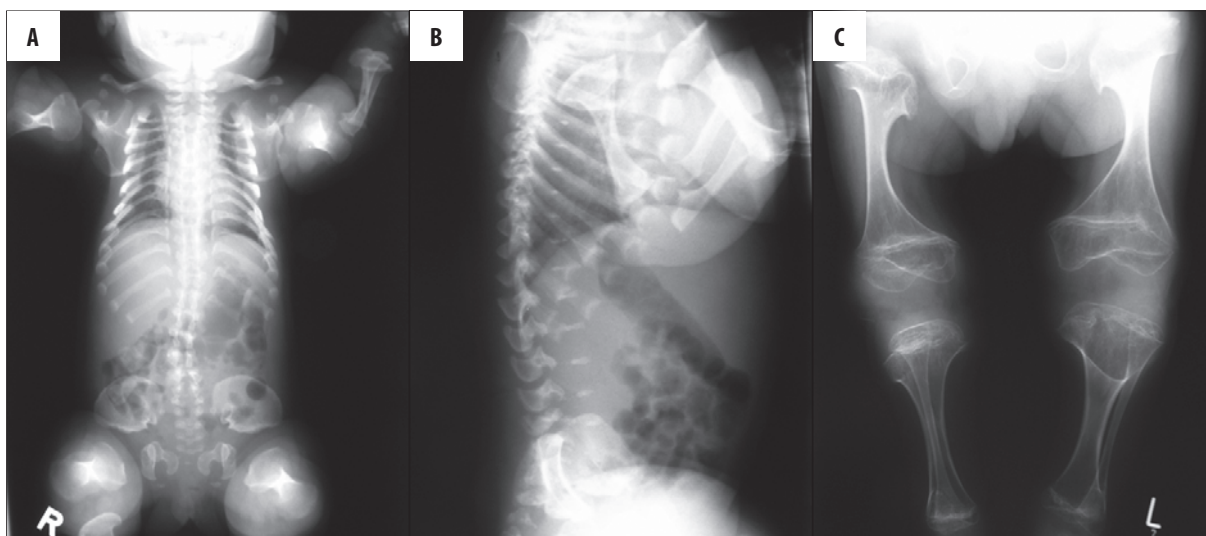


Figure 3. MD Case 3. A&B Newborn. (A) Long trunk. Short extremities widened at the ends. Minor scoliosis is already present in the lumbar spine. Short sacro-iliac notches. Narrow chest. (B) Wafer thin vertebral bodies. (C) At age 3y. Shortened tubular bones with flared metaphyses. Inferiorly directed lesser trochanter.

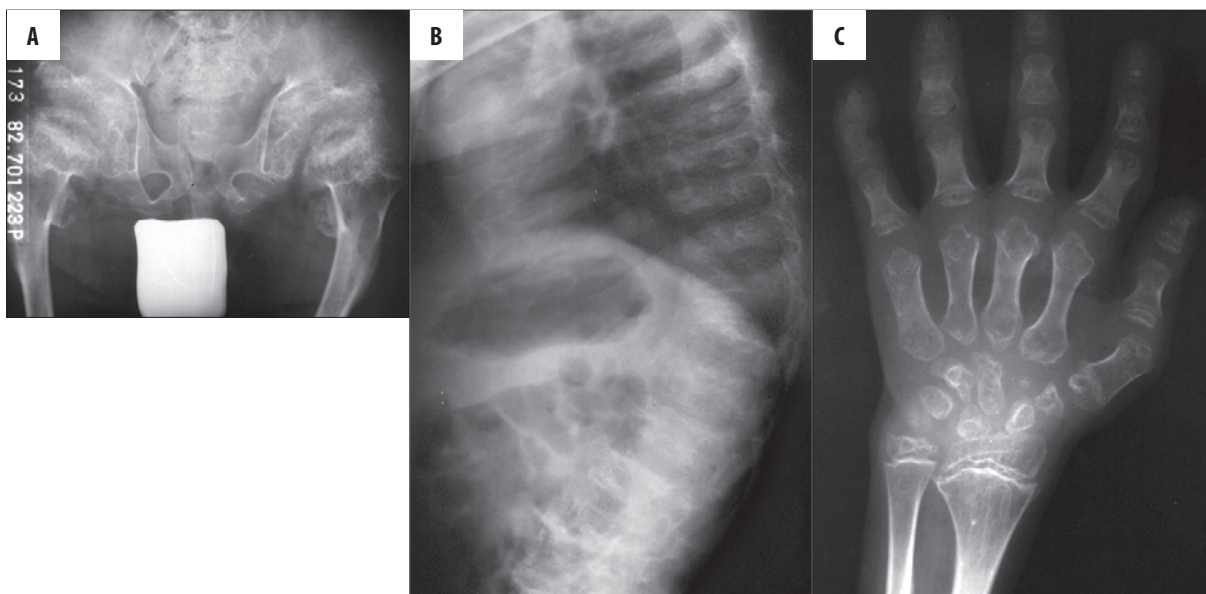


Figure 4. MD CASE 4. At age 12 years. (A) Hypoplasia of the bodies of the iliac bones. Inferiorly directed lesser trochanters. (B) Severe platyspondyly. (C) Severe epi-metaphyseal involvement with flared metaphyses. Hypoplastic/dysplastic carpal bones.

Major radiographic changes are that of a distinctive spondylo-epi-metaphyseal dysplasia. Platyspondyly with kyphosis or kyphoscoliosis is the major feature in the spine. The appearance of the epiphyses might be delayed but when they appear they are small, and may be flattened and irregular in outline. The metaphyses are widened with mushroom appearance in severely affected patients. Marked shortening of the sacro-iliac notches and crescent shaped iliac wings are distinctive features in the pelvis (Figures 3–6).

The radiographic examination is important for several reasons. It not only confirms or excludes the clinical diagnosis but also recognizes the disease in unsuspected cases. Patients with incomplete and minor radiographic expression of MD do occur (MD variants) (Patient 5 – Figure 5).

Until the DNA defect of MD is detected we are not sure if they represent minor forms of MD or a different entity.

The differential diagnosis of MD is with other spondylo-epi-metaphyseal dysplasias. None of them shows such severe platyspondyly with kyphoscoliosis and mushroom shaped metaphyses.

MD is an extremely difficult disorder to handle as kyphoscoliosis progresses in spite of orthrotic treatment. Equally unpredictable is the surgical treatment followed by complications associated with operations on abnormal chondro-osseous tissue [5].

The gene and chromosomal location of MD is unknown. MD is probably the result of a dominant mutation or paren-

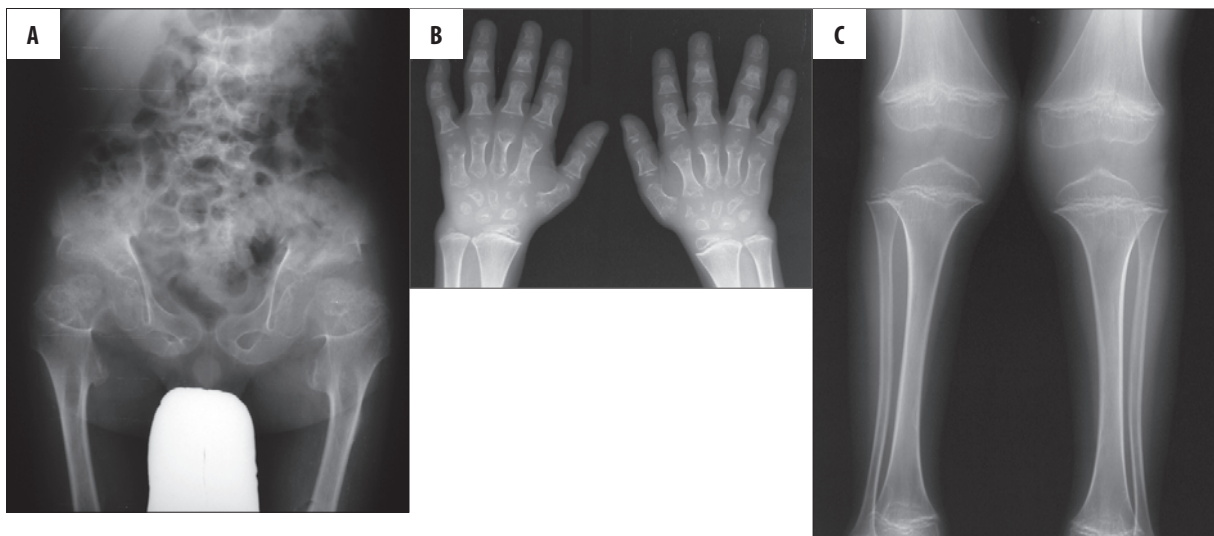


Figure 5. MD variant. Case 6. At age 7 years. (A) Hypoplasia of the bodies of the iliac bones. Minor flaring of the metaphyses. Inferiorly directed lesser trochanters. (B) Short metacarpals and phalanges with flared ends. Hypoplastic/dysplastic carpal bones. (C) The tubular bones are little affected and flaring of the metaphyses is of a minor degree.



Figure 6. MD CASE 7 At age 3 months. Diagnostic features of MD are present on this lower extremities X-ray.

tal germline mosaicism. Recessive mode of inheritance has also been suggested.

Conclusions

Abnormal measurements in the newborn are an indication for thorough, perspicacious clinical examination. In doubtful cases five basic radiographs - lateral skull, lateral spine, pelvis, one hand and legs - should be considered to exclude or identify a bone dysplasia.

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